Genetic Testing



Our bodies are made up of trillions of tiny pieces. These pieces are called cells. Inside every cell is an instruction manual that tells the cell what to do. This instruction manual is called DNA. Some people have health problems because they have misspellings in their DNA. These misspellings are sometimes called variants. Genetic testing is the process of analyzing someone's DNA.

What are the Benefits?

- Finding DNA misspellings can help us discover other health problems.
- Some genetic diseases have specific treatments.
- Genetic testing can find the risk for future children's possible health problems.

How much does it cost?

- Covered by many insurances.
- Some insurance companies will require a prior authorization before they will approve payment for the test.
- When the cost to the patient is estimated to be:
  - less than $100 - test is often started.
  - higher than $100 - you may be contacted to see if you want to pay or cancel the test.

How long does it take?

- 4-8 weeks

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Possible Results

- **Positive:** a misspelling was found that explains a health problem.
- **Negative:** no misspelling was found; risk of disease is lower.
  - Many new types of testing developed yearly. It is possible there are misspellings the testing did not find.
- **Variant of Uncertain Significance (VUS):** Test found a misspelling it had not seen before. Your provider will research the VUS and let you know if they believe it to be important or not.

What's Next?

1. Sample Collection:

We will need to collect a sample to analyze you or your child's DNA. Most genetic testing can be performed on a cheek swab, but sometimes it will require a blood draw. Your clinician may swab at the appointment or send a kit to your house.

2. Call from Genetic Counselor:

A genetic counselor may contact you after the appointment to discuss changes to the genetic test. Either to get additional samples from a parent or sibling or discuss insurance.

3. Call from Laboratory:

Some labs will contact you if your cost will be more than $100. Be sure to talk to them if they call.

4. Wait for Results:

There are many different genetic tests. Typically, you will get results within 3-8 weeks. The most common test takes between 6-8 weeks.

5. Discuss Results with Your Clinician:

Once the results are received, your clinician will either call or request to discuss them with you in person at the next appointment.

Example: Emily was recently diagnosed with developmental delay. Her doctor ordered a genetic test called whole exome sequencing. This test found a variant in the PTPN11 gene, which is associated with a disease called Noonan syndrome. Noonan syndrome can cause developmental delay AND a heart disease called cardiomyopathy. Because of this genetic test, Emily can now be properly screened for heart disease.

If you have questions about genetic testing, please talk to your healthcare clinician.